Apert syndrome

Apert syndrome is a genetic disorder characterized by the premature fusion of certain skull bones (craniosynostosis). This early fusion prevents the skull from growing normally and affects the shape of the head and face. In addition, a varied number of fingers and toes are fused together (syndactyly).

Many of the characteristic facial features of Apert syndrome result from the premature fusion of the skull bones. The head is unable to grow normally, which leads to a sunken appearance in the middle of the face, bulging and wide-set eyes, a beaked nose, and an underdeveloped upper jaw leading to crowded teeth and other dental problems. Shallow eye sockets can cause vision problems. Early fusion of the skull bones also affects the development of the brain, which can disrupt intellectual development. Cognitive abilities in people with Apert syndrome range from normal to mild or moderate intellectual disability.

Individuals with Apert syndrome have webbed or fused fingers and toes. The severity of the fusion varies; at a minimum, three digits on each hand and foot are fused together. In the most severe cases, all of the fingers and toes are fused. Less commonly, people with this condition may have extra fingers or toes (polydactyly). Additional signs and symptoms of Apert syndrome can include hearing loss, unusually heavy sweating (hyperhidrosis), oily skin with severe acne, patches of missing hair in the eyebrows, fusion of spinal bones in the neck (cervical vertebrae), and recurrent ear infections that may be associated with an opening in the roof of the mouth (a cleft palate).

Frequency

Apert syndrome affects an estimated 1 in 65,000 to 88,000 newborns.

Genetic Changes

Mutations in the *FGFR2* gene cause Apert syndrome. This gene produces a protein called fibroblast growth factor receptor 2. Among its multiple functions, this protein signals immature cells to become bone cells during embryonic development. A mutation in a specific part of the *FGFR2* gene alters the protein and causes prolonged signaling, which can promote the premature fusion of bones in the skull, hands, and feet.

Inheritance Pattern

Apert syndrome is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Almost all cases of Apert syndrome result from new mutations in the gene, and occur in people with no

history of the disorder in their family. Individuals with Apert syndrome, however, can pass along the condition to the next generation.

Other Names for This Condition

- Acrocephalosyndactyly (Apert)

Diagnosis & Management

These resources address the diagnosis or management of Apert syndrome:

- GeneReview: FGFR-Related Craniosynostosis Syndromes
  https://www.ncbi.nlm.nih.gov/books/NBK1455
- Genetic Testing Registry: Acrocephalosyndactyly type I
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0001193/
- MedlinePlus Encyclopedia: Apert syndrome
  https://medlineplus.gov/ency/article/001581.htm
- MedlinePlus Encyclopedia: Webbing of the fingers or toes
  https://medlineplus.gov/ency/article/003289.htm

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Apert syndrome
  https://medlineplus.gov/ency/article/001581.htm
- Encyclopedia: Webbing of the fingers or toes
  https://medlineplus.gov/ency/article/003289.htm
- Health Topic: Craniofacial Abnormalities
  https://medlineplus.gov/craniofacialabnormalities.html

Genetic and Rare Diseases Information Center

- Apert syndrome
  https://rarediseases.info.nih.gov/diseases/5833/apert-syndrome

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Craniosynostosis Information Page
  https://www.ninds.nih.gov/Disorders/All-Disorders/Craniosynostosis-Information-Page

Educational Resources

- Boston Children's Hospital
  http://www.childrenshospital.org/conditions-and-treatments/conditions/apert-syndrome
- Collaboration for Craniofacial Development and Disorders, Johns Hopkins University
  http://www.hopkinsmedicine.org/neurology_neurosurgery/centers_clinics/pediatric_neurosurgery/conditions/craniosynostosis/
- Disease InfoSearch: Apert Syndrome
  http://www.diseaseinfosearch.org/Apert+Syndrome/544
- MalaCards: apert syndrome
  http://www.malacards.org/card/apert_syndrome
- Orphanet: Apert syndrome
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=87
- Orphanet: Craniosynostosis
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1531
- Seattle Children's Hospital and Regional Medical Center
  http://www.seattlechildrens.org/medical-conditions/chromosomal-genetic-conditions/apert-syndrome/
- The Craniofacial Center, Dallas, Texas
  http://thecraniofacialcenter.com/apert.html
- U.C. Davis Children's Hospital
  http://www.ucdmc.ucdavis.edu/children/clinical_services/cleft_craniofacial/anomalies/apert.html

Patient Support and Advocacy Resources

- AmeriFace
  http://www.ameriface.org/
- Children's Craniofacial Association
  http://www.ccakids.com
- National Organization for Rare Disorders (NORD)
  http://rarediseases.org/rare-diseases/apert-syndrome/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/apert.html

GeneReviews

- FGFR-Related Craniosynostosis Syndromes
  https://www.ncbi.nlm.nih.gov/books/NBK1455

Genetic Testing Registry

- Acrocephalosyndactyly type I
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0001193/

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22Acrocephalosyndactylia%22+OR+%22Apert+syndrome%22+OR+%22Craniosynostoses%22

Scientific articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Acrocephalosyndactylia%5BMAJR%5D%29+AND+%28%28apert+syndrome%5BTIAB%5D%29+OR+%28acrocephalosyndactyly%5BTIAB%5D%29+OR+%28acrocephaly%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

OMIM

- APERT SYNDROME
  http://omim.org/entry/101200

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